



CATSPER1-related nonsyndromic male infertility

CATSPER1-related nonsyndromic male infertility is a condition that affects the function of sperm, leading to an inability to father children. Males with this condition produce sperm that have decreased movement (motility). Affected men may also produce a smaller than usual number of sperm cells or sperm cells that are abnormally shaped. Men with *CATSPER1*-related nonsyndromic male infertility do not have any other symptoms related to this condition.

Frequency

The prevalence of *CATSPER1*-related nonsyndromic male infertility is unknown.

Genetic Changes

Mutations in the *CATSPER1* gene cause *CATSPER1*-related nonsyndromic male infertility. The *CATSPER1* gene provides instructions for producing a protein that is found in the tail of sperm cells. The *CATSPER1* protein is involved in the movement of the sperm tail, which propels the sperm forward and is required for sperm cells to push through the outside membrane of the egg cell during fertilization.

CATSPER1 gene mutations result in the production of a *CATSPER1* protein that may be altered, nonfunctional, or quickly broken down (degraded) by the cell. Sperm cells missing a functional *CATSPER1* protein have decreased motion in their tails and move more slowly than normal. Sperm cells lacking functional *CATSPER1* protein cannot push through the outside membrane of the egg cell. As a result, sperm cells cannot reach the inside of the egg cell to achieve fertilization.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show symptoms of the condition.

Males with two *CATSPER1* gene mutations in each cell have *CATSPER1*-related nonsyndromic male infertility. Females with two *CATSPER1* gene mutations in each cell have no symptoms because the mutations only affect sperm function, and women do not produce sperm.

Other Names for This Condition

- CATSPER-related nonsyndromic male infertility
- CATSPER1-related male infertility

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: CATSPER-Related Male Infertility
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2751811/>

Other Diagnosis and Management Resources

- Cleveland Clinic: Male Infertility
<http://my.clevelandclinic.org/health/articles/male-infertility>
- GeneReview: CATSPER-Related Male Infertility
<https://www.ncbi.nlm.nih.gov/books/NBK22925>
- MedlinePlus Health Topic: Assisted Reproductive Technology
<https://medlineplus.gov/assistedreproductivetechnology.html>
- RESOLVE: The National Infertility Association: Semen Analysis
<http://www.resolve.org/about-infertility/male-workup/the-semen-analysis.html>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Semen Analysis
<https://medlineplus.gov/ency/article/003627.htm>
- Health Topic: Assisted Reproductive Technology
<https://medlineplus.gov/assistedreproductivetechnology.html>
- Health Topic: Male Infertility
<https://medlineplus.gov/maleinfertility.html>

Educational Resources

- Cleveland Clinic: Male Infertility
<http://my.clevelandclinic.org/health/articles/male-infertility>
- Disease InfoSearch: CATSPER-Related Male Infertility
<http://www.diseaseinfosearch.org/CATSPER-Related+Male+Infertility/7928>
- MalaCards: catsper1-related nonsyndromic male infertility
http://www.malacards.org/card/catsper1_related_nonsyndromic_male_infertility
- Merck Manual Professional Version: Sperm Disorders
<http://www.merckmanuals.com/professional/gynecology-and-obstetrics/infertility/sperm-disorders>

Patient Support and Advocacy Resources

- RESOLVE: The National Infertility Association
<http://www.resolve.org/>
- University of Kansas Medical Center Resource List: Infertility
<http://www.kumc.edu/gec/support/inferti.html>

GeneReviews

- CATSPER-Related Male Infertility
<https://www.ncbi.nlm.nih.gov/books/NBK22925>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22CATSPER1-related+male+infertility%22+OR+%22Infertility%2C+Male%22+OR+%22Male+Infertility%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CATSPER1%5BALL%5D%29+AND+%28infertility%5BALL%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- SPERMATOGENIC FAILURE 7
<http://omim.org/entry/612997>

Sources for This Summary

- Avenarius MR, Hildebrand MS, Zhang Y, Meyer NC, Smith LL, Kahrizi K, Najmabadi H, Smith RJ. Human male infertility caused by mutations in the CATSPER1 channel protein. *Am J Hum Genet.* 2009 Apr;84(4):505-10. doi: 10.1016/j.ajhg.2009.03.004. Epub 2009 Apr 2.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19344877>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2667975/>
- GeneReview: CATSPER-Related Male Infertility
<https://www.ncbi.nlm.nih.gov/books/NBK22925>
- Li HG, Liao AH, Ding XF, Zhou H, Xiong CL. The expression and significance of CATSPER1 in human testis and ejaculated spermatozoa. *Asian J Androl.* 2006 May;8(3):301-6.
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